correlation resulting in misinterpretation of chromatographic profiles. This is especially important when there is the presence of metabolites associated with OA, such as MMA, GA, PA, and IVA, considering that some of them are present in normal profiles and secondary to other clinical conditions. Taking this into account, diagnostic utility of some metabolites depended on patient's clinical condition, age, and presence of biochemical-related metabolites. **Conclusion:** The GC-MS qualitative analysis results must be interpreted according to the patient's clinical context, and for some cases, a long-term follow-up and even molecular studies may be required to define diagnosis.

092 - Ornithine Transcarbamylase Deficiency: Identification of Mutations, Computational Validation, and Phenotypic Correlation in Argentinian Patients

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Introduction: Ornithine transcarbamylase deficiency (OTCD; *Online Mendelian Inheritance in Man* [OMIM] 311250) is an urea cycle defect with X-linked inheritance. In hemizygous males, neonatal or late onset depends on the degree of residual enzymatic activity. In heterozygous females, symptom presentation depends on X-chromosome inactivation. Mutation identification in OTC gene allows diagnostic confirmation and carrier detection. **Objective:** To identify mutations causing OTCD in Argentinian patients, to validate those changes, and to correlate them with phenotype. **Methods:** A total of 11 patients belonging to 8 families, 6 male patients, 2 with severe presentation and death during the neonatal period and 4 with late-onset (0.5-10 years) and 5 symptomatic women (0.8 to 4 years), 3 of them died and were diagnosed with OTCD. Molecular analysis of OTC gene was performed by polymerase chain reaction/multiplex ligation-dependent probe amplification/Single-strand conformation polymorphism analysis and/or sequencing, and missense changes validation was made using computational methods, PolyPhen, SIFT, and PopMusic 2.0. **Results:** We identified mutations in all patients; 2 were not previously described: 1 of splicing (c.540+1G>A) and a deletion (delExon 2-10) and 6 were already reported: 1 of splicing (c.216+1G>A) and 5 missense (p.Arg129His, p.Leu151Arg, p.Thr178Met, p.Ala208Thr, and p.Arg277Trp). Result validation was consistent with the applied computational programs and the patients' presentation form. **Conclusion:** This analysis provides a better understanding of alterations responsible for the phenotypic expression. This work expands carrier detection capability allowing appropriate genetic counseling. Early detection of patients with OTCD is essential to reduce morbidity and mortality in affected individuals.

093 - Overgowth in the First Year of Life: An Early Sign of Mucopolysaccharidosis

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Introduction: Child's growth is one of the best indicators of overall health, and it can be impacted by chronic diseases such as mucopolysaccharidosis (MPS). Paradoxically, growth in the first years of life in some types of MPS tends to be accelerated. Patients with MPS appear healthy at birth. The initial signs and symptoms in most patients with MPS have been identified after 2 years, which delays diagnoses. **Objective:** The purpose of this study was to analyze growth in the first year of life in patients with MPS VI, IVA and IIIC as an early sign of the disease. **Methodology:** A total of 8 patients (4 boys, 4 girls), 6 with MPS IVA (4 boys, 2 girls), 1 MPS VI (girl), and 1 MPS IIIC (girl) with at least 5 length measurements in the first year of life, were identified. The length data were obtained from their medical records. The patients' data were plotted on the World Health Organization growth chart including diverse ethnic backgrounds. **Results:** Length in girls, the curve was above P97 in 2 of 4 patients (MPSIVA, MPSVI), between P95 and P97 in 1 of 4 (MPSIIIC), and between P50 and P85 in 1/4 (MPSIVA). Length in boys: of 4 patients with MPSIVA the curve was above the P97 in 2 of 4 patients, between P95 and P97 1 of 4 patients, and between P85 and P95 in 1 of 4 patients. **Conclusion/Discussion:** The MPS is a diagnosis that should be taken into consideration when differentiating the causes of overgrowth in the first year of life in order to improve the early diagnosis and treatment of MPS.

094 - Overview of the First 3 Years of Operation (2010-2012) of the Niemann-Pick Type C Brazil Network

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Niemann-Pick type C is an autosomal recessive inborn error of cholesterol trafficking, characterized by the storage of cholesterol inside the lysosomes, which leads to a wide range of clinical manifestations, usually involving the central nervous system. As there is a specific treatment already approved and several therapeutic strategies in development, diagnosis is